Opis choroby *

Definicja

An exceedingly rare, autosomal recessive immune disease characterized by thumb aplasia, short stature with skeletal abnormalities, and combined immunodeficiency described in three sibships from two possibly related families. The skeletal abnormalities included unfused olecranon and the immunodeficiency manifested with severe chickenpox and chronic candidiasis. No new cases have been reported since 1978.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2951

Kod OMIM
274190

Kod ICD10
D82.8

Kod ICD11
4A01.1Y

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*Źródło

orphanet